CASE REPORT

Transperineal aggressive angiomyxoma

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SUMMARY

A 45-year-old woman with a history of total hysterectomy with adnexal preservation for uterine leiomyomas presented to our hospital with a right gluteal palpable mass, which she first noticed 6 months before and had progressively enlarged since then.

Radiological studies revealed a 14 cm lesion with translevator growth that displaced rather than invaded adjacent structures, with a peculiar whorled pattern on T2-weighted MRI, which enhanced following gadolinium administration. CT-guided biopsy was performed, and in conjunction with imaging features the diagnosis of an aggressive angiomyxoma was assumed and confirmed following surgical excision.

BACKGROUND

Aggressive angiomyxoma is a rare benign soft-tissue neoplasm most frequently found in premenopausal women. As it generally arises from the perineum and the pelvic retroperitoneum, it creates some meaningful differential diagnosis issues as the therapeutic options, including surgical protocol, and outcomes differ considerably according to the histology of the lesion.

We intend to emphasise the imaging features that prompted the preoperative diagnosis, and discuss the histological features, the differential diagnosis and the chosen course of treatment.

CASE PRESENTATION

A 45-year-old female was referred to the outpatient surgical clinic of our oncological institution due to a right gluteal swelling, progressively enlarging over the past 6 months, associated with perineal pain. The patient had undergone a total abdominal hysterectomy 10 years earlier for uterine leiomyomas, with adnexal preservation.

Physical examination revealed a soft right pararectal lesion extending into the right gluteus. There were no intrinsic vaginal and rectal lesions, but an extrinsic bulging of these organs was felt on examination.

Blood work and tumorous markers were unremarkable.

INVESTIGATIONS

Pelvic CT and MRI were obtained. Contrast-enhanced CT revealed a predominantly solid, heterogeneously enhancing lesion, measuring 5.5×6.5×14 cm, extending from the fat of the right ischioanal fossa to the level of the vaginal dome, displacing the rectum laterally and the vagina anteriorly, without signs of bowel obstruction. Even though these structures did not seem to be invaded, there was no identifiable fat plane between the lesion and the vagina (figure 1).

The full extent of the lesion was better depicted using MRI, which revealed a lesion with low-signal intensity on T1-weighted images and heterogeneous high-signal intensity on T2-weighted images with multiple hypointense linear strands arranged into a whorled pattern on axial images and into a laminated pattern on sagittal images, roughly showing an 'hourglass' shape due to the constriction caused posteriorly by the coccyx and the growth through a breach on the levator ani muscles. The whorled appearance was further demonstrated following contrast enhancement with gadolinium. Neither the right levator ani muscles nor adjacent viscera showed signs of invasion, indicating that these were only displaced as depicted on CT. Both ovaries were identified, clearly demarcated from the lesion (figure 2).

The remainder of the systemic investigation excluded signs of metastasis.

CT-guided core needle biopsy was performed and the presumptive diagnosis of an aggressive angiomyxoma was confirmed; however, low-grade sarcoma could not be excluded.

DIFFERENTIAL DIAGNOSIS

Clinically, aggressive angiomyxomas are understandably mistaken for lipomas, Bartholin duct cysts, rectoceles, hernias or even relatively prevalent malignant pelvic floor tumours, like squamous cell carcinomas or soft-tissue sarcomas.[1–4]

Radiologically though, most of these lesions are promptly ruled out, and even if imaging features are not pathognomonic for aggressive angiomyxomas they can be very suggestive, as was with the case presented here. The differential diagnosis should include angiomyofibroblastoma, soft-tissue sarcomas, pelvic myxoma and solitary fibrous tumour, despite the fact that all these lesions lack laminated/whorled pattern on T2-weighted images associated with aggressive angiomyxomas.[1 5 6]

Angiomyofibroblastoma is a rare benign mesenchymal tumour, with much the same gender, age group and site of origin predilection as aggressive angiomyxoma. Its radiological appearance is also rather similar to that of aggressive angiomyxomas, making these two entities virtually indistinguishable if the whorled pattern is not present or there is no translevator growth, as in aggressive angiomyxomas.[7]

The group of soft-tissue sarcomas includes lesions like the rhabdomyosarcoma, liposarcoma and undifferentiated pleomorphic sarcoma, formerly known as 'malignant fibrous histiocytoma'.[8] Rhabdomyosarcomas occur in much younger patients. Imaging features of liposarcomas vary according

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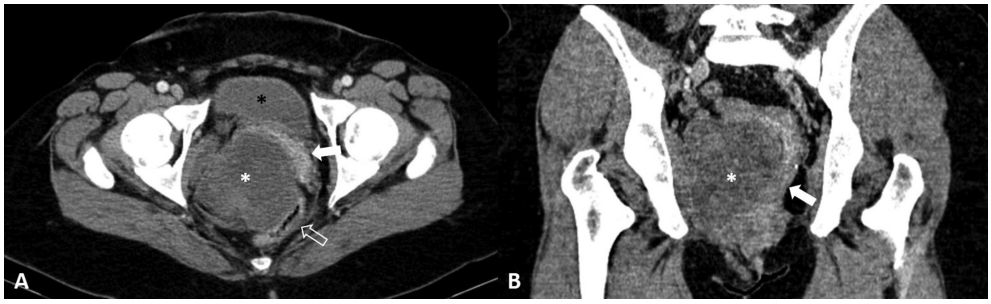

Figure 1 Axial contrast-enhanced CT (**A**) and coronal reformatted (**B**) images showing a heterogeneous lesion (white asterisk), extending from the right ischioanal fossa to the vaginal dome. The rectum is displaced laterally (white open arrow) and the vagina (white arrow) anteriorly and laterally, with no fat plane identifiable with this structure. Black asterisk, bladder.

to their grade, but they either contain fat or are locally invasive. Undifferentiated pleomorphic sarcomas are heterogeneous lesions, especially on MRI due to haemorrhage, necrosis or calcifications, and their origin on adjacent muscles might be identifiable.[6 9]

Myxomas are also benign mesenchymal soft-tissue tumours found on the heart, skeletal muscle, somatic soft tissues and gastrointestinal or genitourinary tract. On CT, they are well-circumscribed low-attenuation lesions. On MRI, they reveal homogeneous low-signal intensity on T1-weighted images and high-signal intensity on T2-weighted images. Cystic areas are usually present and the enhancement pattern is variable.[6]

Solitary fibrous tumours very rarely occur in the perineum. On imaging studies they are hypervascular and also displace rather than invade adjacent structures. However, especially when large, they tend to be heterogeneous lesions, with areas of high-signal intensity on T1-weighted images and multiple flow voids on T2-weighted images, due to haemorrhagic areas and large feeding vessels, respectively.[6]

TREATMENT

The patient underwent surgical resection of the lesion, employing a simultaneous perineal and abdominal approach which achieved an en-block resection. The postoperative period was uneventful.

OUTCOME AND FOLLOW-UP

Gross examination of the surgical specimen revealed an anfractuous bland tumour, focally coincident with the resection margin.

Microscopically, it was an infiltrative paucicellular tumour, composed of cells with inconspicuous cytoplasm and small bland ovoid to short spindle-shaped nuclei, embedded in an abundant myxoid matrix. Mitotic activity and nuclear atypia were absent. Numerous blood vessels of varying calibre, ranging from thin-walled, capillary-like vessels of medium calibre, to large vessels with thick muscular walls, were present. The infiltrative edge entrapped adipose tissue and skeletal muscle at the periphery. Phenotypically, the cells were positive for oestrogen and progesterone receptors (figure 3).

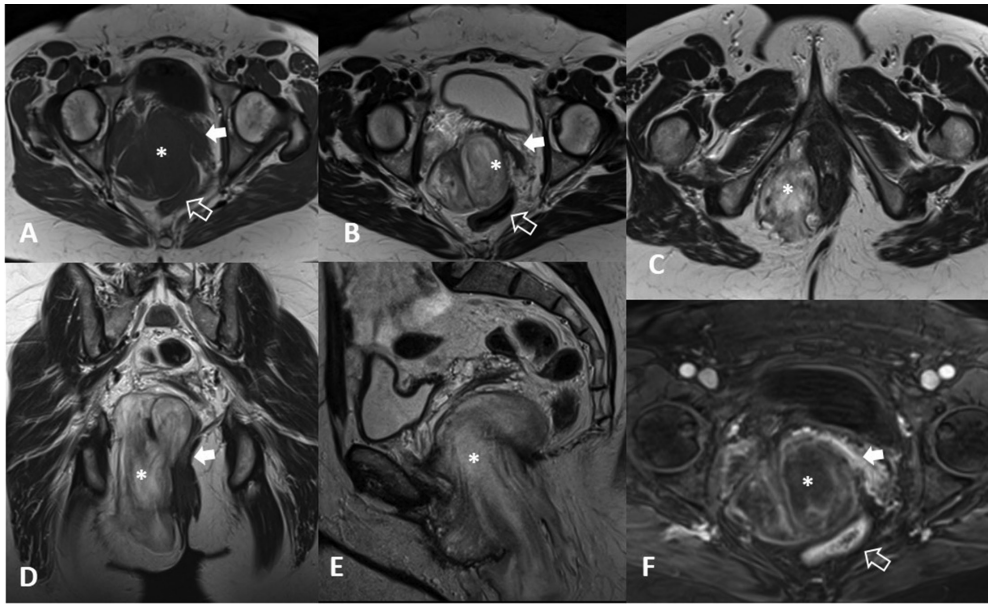

Figure 2 Axial T1-weighted (**A**), axial T2-weighted (**B,C**), coronal T2-weighted (**D**), sagittal T2-weighted (**E**) and axial T1-weighted fat-saturated gadolinium-enhanced (**F**) sequences showing a homogeneously hypointense lesion (white asterisk) on T1-weighted sequences (**A**), with heterogeneous high-signal intensity on T2-weighted sequences (**B–E**). The lesion enhances intensely (**F**). The laminated appearance given by the multiple hypointense strands is better depicted on the coronal (**D**) and sagittal (**E**) planes, which on the axial plane accounts for the whorled appearance (**B**). The 'hourglass' shape is seen on (E). No invasion of adjacent structures is present. White open arrow, rectum; white arrow, vagina.

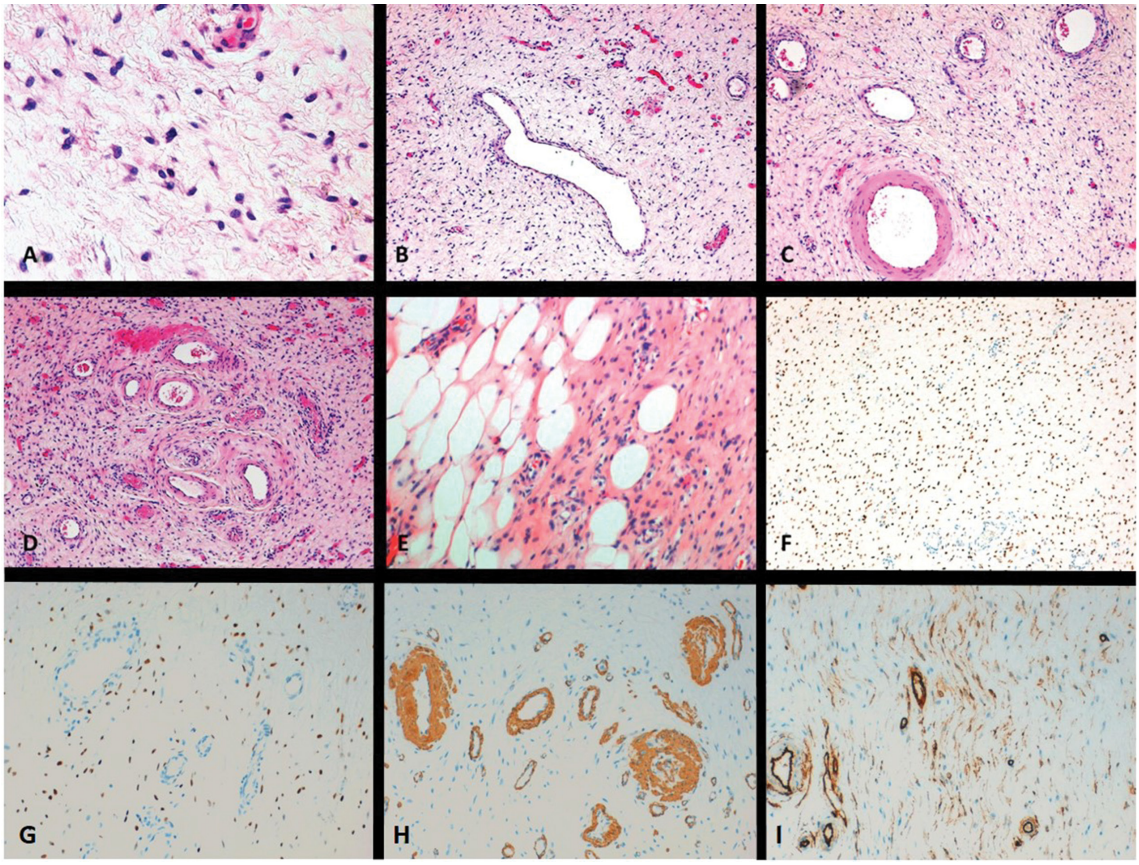

Figure 3 The tumour is composed of small spindle and stellate fibroblasts with no atypia, within a myxoid stroma (A, 400x), with collagen fibres and prominent, dilated, thick-walled vessels (B,D, 100x), which may be hyalinised with a prominent vascular smooth muscle layer (C, 100x). Peripheral infiltrative margins with extension into surrounding adipose tissue can be seen (E, 100x). The cells are positive for oestrogen (F, 200x) and progesterone receptors (G, 200x), focally positive for CD34 (H, 200x) and negative for smooth muscle actin (I, 200x).

The patient has been under follow-up for 1 year now, and remains asymptomatic and with no radiological evidence of recurrence.

DISCUSSION

Aggressive angiomyxomas are rare soft-tissue neoplasms. Although they have a predilection for pelvic and perineal regions, there are reported cases described in the abdomen and kidney.[10 11] It occurs predominantly in premenopausal women, with a female to male ratio of 6:1, and between the third and sixth decades with a peak incidence in the fourth decade. There are no known associated aetiological factors.[10 12]

It is classified as a benign tumour with locally aggressive behaviour owning to its infiltrative nature, displacing rather than invading adjacent structures, even though there are a few rare reports of bladder, rectal and bone invasion.[11 12] This infiltrative nature makes complete excision with free margins somewhat difficult, explaining the recurrence rates greater than 35%.[4 11] To our knowledge only two cases of metastatic aggressive angiomyxoma have been described.[3 13]

The majority of patients present with an indolent growing asymptomatic mass. Therefore these lesions can reach large dimensions on presentation. When present, symptoms relate to the mass effect and frequently include pelvic pressure, pain, dyspareunia or bowel obstruction.[1 4 11]

Due to their size and typical translevator growth, clinical examination does not take into account the pelvic and retroperitoneal components of the lesions, underestimating its size. Therefore radiological characterisation is of utmost importance to surgical planning.[1 3 4] MRI will provide the most accurate data for both characterisation and extent evaluation, crucial for surgical planning. Aggressive angiomyxomas are isointense to muscle on T1-weighted sequences and hyperintense on T2-weighted sequences, with heterogeneous intense contrast enhancement. The laminated/whorled appearance on T2-weighted sequences and enhancement in postcontrast studies is highly suggestive of the diagnosis. It is reported to be present in at least 83% of cases.[11]

Surgery is the treatment of choice for symptomatic primary or recurrent disease.[1 3 4 11] Gonadotropin-releasing hormone agonists have been successfully used mainly on recurrent disease, whenever the tumour is positive for oestrogen/progesterone receptors.[1 14] Complete remissions have been reported, and even if not achieved, the downsizing of the tumour might be invaluable for reducing surgical morbidity.[4 15] The role of adjuvant hormonotherapy in reducing recurrence rates is yet to be proven.[15] Radiotherapy and chemotherapy do not have a role on the management of these lesions due to their low mitotic index.[1] Taking this into account, the decision was made to only use hormonotherapy in case of recurrence, which has not happened during the first year of follow-up.

Learning points

- Aggressive angiomyxoma is a rare benign soft-tissue neoplasm most frequently found in premenopausal women, with a predilection for the perineum and pelvis.
- Clinically it is frequently mistaken for other more prevalent perineal lesions and, due to the translevator growth, its extension is largely underestimated, which accounts for the high local recurrence rates.
- Radiological studies, especially MRI, are essential for determining the extension and characterise this tumour. The translevator growth and the laminated/whorled appearance on T2-weighted sequences and enhancement in postcontrast studies are highly suggestive of the diagnosis.
- Surgical excision is the treatment of choice for both primary and recurrent disease, and hormonotherapy may be used for downsizing recurrent disease.

Contributors PP did the bibliographical research and wrote the manuscript. EMA was responsible for editing the manuscript. IR provided the histological images of the tumour, with respective pathological analysis and diagnosis. TMC revised the manuscript and gave it final approval.

Competing interests None declared.

Patient consent Obtained.

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